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A rare case report of persistent mullerian duct syndrome: Incidental finding in a case of ascending colon carcinoma

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Abstract
Persistent mullerian duct syndrome (PMDS) is usually an accidental finding during any surgery in male patients presenting with undescended testis or absent testis. It is caused by a defect in synthesis or action of mullerian inhibiting substance system.
Intraoperatively mullerian remnants consisting of an infantile uterus and fallopian tubes are usually found. We report a case of 25 years old male patient presenting with right side abdominal lump and left side absent testis.

Keywords: Persistent mullerian duct syndrome, undescended or absent testis, mullerian inhibiting substance

Introduction
Intersexual disorders are very important clinical issues with their different aspects relating to diagnosis, treatment and sex of rearing [2]. Persistent Mullerian duct syndrome is a rare form of internal male pseudo hermaphroditism can occur due to mutation in MIS and MISR-2 Gene has autosomal recessive transmission [1]. They are broadly classified into disorders associated with an abnormal chromosome constitution and normal chromosome constitution. Patients are phenotypically male and usually present wen young with unilateral or bilateral cryptorchid testis and inguinal hernia into which prolapses an infantile uterus and fallopian tubes [3]. Familial cases have been reported with a probability of sex-limited autosomal recessive or X-linked recessive inheritance. An incidence of PMDS in identical twins has also been reported [4]. PMDS is often misdiagnosed due to a lack of familiarity with the condition. A review of the literature showed only about few case reports of this including familial cases.

Case Report
A 25 year old male presented at OPD of Smt. SMS Multispeciality Hospital with complain of right side abdominal lump since 1 year, per rectal bleeding since 6 month. Patient having a short stature and weight is 31kg. Poor secondary sexual characterisitics. There is no history of backpain, coughing, Hemoptysis. On clinical finding there is a aprox 5 * 4 cm ill-defined lump on right lumbar region. Liver is not enlarged and left side absent testis with hypospadias.

CECT abdomen with pelvis: Suggestive of a well-defined, elongated, peripherally enhancing tubular structure at Recto-prostatic space just posterior to urinary bladder on left side; suggest? Seminal vesical cyst and large circumferential wall thickening involving proximal ascending colon? Malignant lesion.

Colonoscopy with biopsy: There is a Noduloproliferative mass seen with luminal narrowing.

Biopsy: Adenocarcinoma, Moderately differentiated.

Surgery: For ascending colon carcinoma: laparotomy and right hemicolectomy, ileo-transverse anastomosis with proximal ileostomy done. Intraoperatively we found tubular

structure over pelvic region on left side. Its look like a rudimentary uterus with fallopian tube. We send it for histopathological examination.

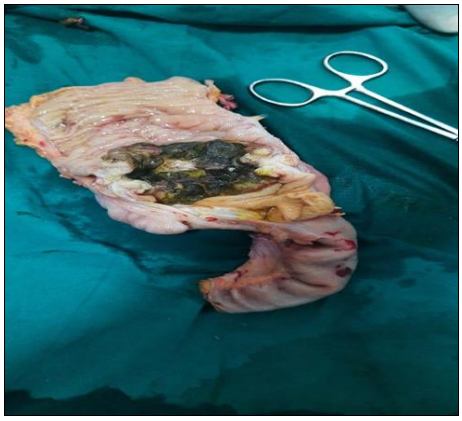

Fig 1: Specimen of ascending colon

Histopathological examination: Section show structure of uterus with atrophic endometrial glands, normal myometrial muscle fibers. Fallopian tubes are identified and show normal histology with fibrosis section from cervix show flattened squamous epithelium and endocervical glands and poorly differentiated Adenocarcinoma of ascending colon.

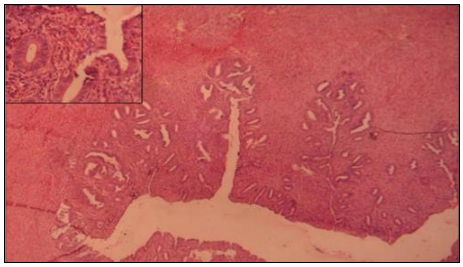

Fig 2: Immature uterus with endometrial lining

Stage: T3N0Mx. All lymphnodes are free from metastasis. We did karyotyping of this patient: (46, X, + Marker chromosome)

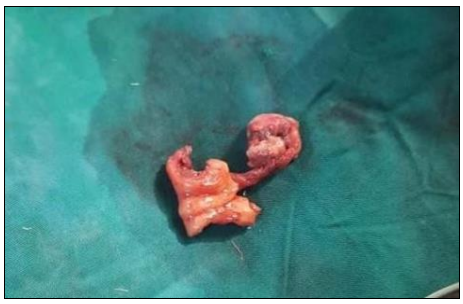

Fig 3: Specimen of rudimentary uterus with fallopian tube

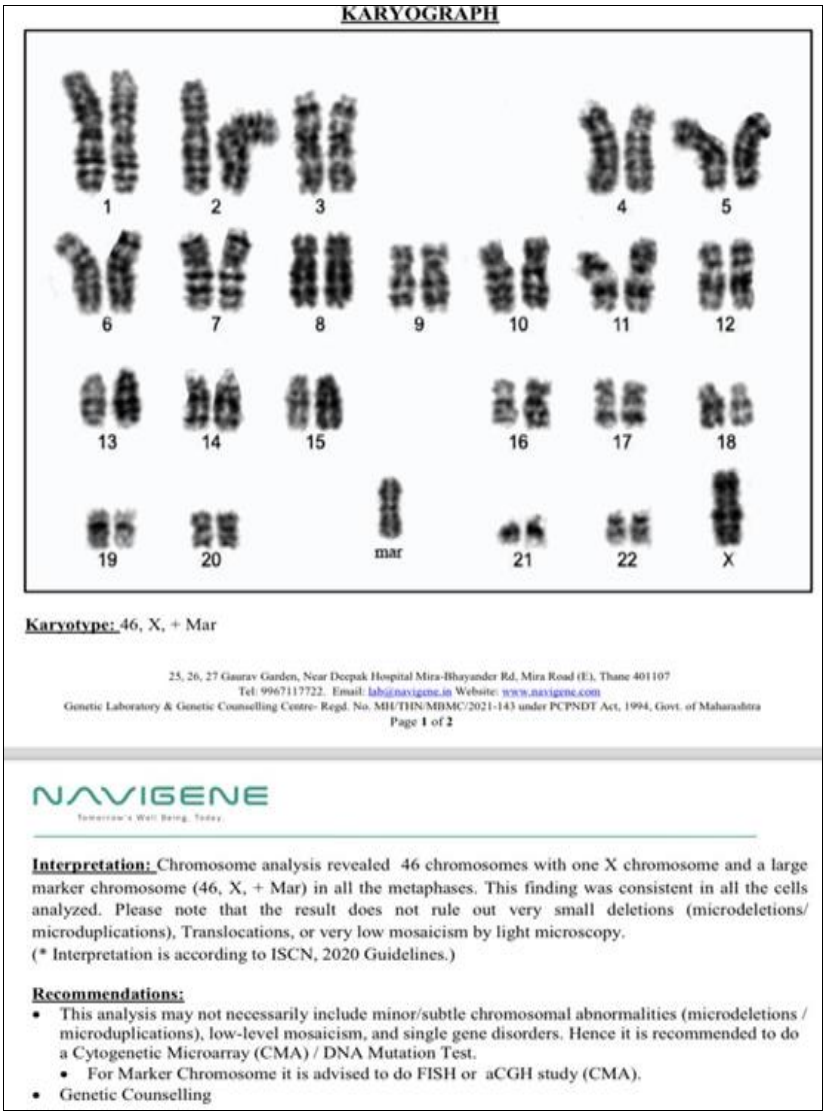

Fig 4: Chromosome Analysis test Report

Discussion

The MIS is a large glycoprotein that Sertoli cells produce early in fetal life. The gene responsible for the substance is on chromosome 19. MIS is also known as an anti mullerian hormone. The primary function of MIS is to cause regression of the mullerian (Paramesonephric) ducts in the male fetus. MIS is first secreted in effective amounts 56-62 days after fertilization, and the process of mullerian regression is normally completed by about day 77, after which mullerian tissue is no longer sensitive to MIS.

Clinically, PMDS cases are divided into three categories [3-5]

- Majority (60-70%) with bilateral intraabdominal testis in apposition analogous to ovaries.
- Smaller group (20-30%) with one testis in scrotum associated contralateral inguinal hernia whose contents are testis, uterus and tubes. (Hernia uteri inguinale)
- Smallest group (10%) where both testis are located in the same hernia sac along with the mullerian structures. (Transverse testicular ectopia-TTE)

Our case did not show any dysplastic changes or malignancy. However there are reported cases with PMDS associated with testicular malignancy [5].

The diagnosis of PMDS is made incidentally during surgical exploration as the mullerian remnants are not palpable on abdominal, rectal or scrotal examination. Intraoperative methods of diagnosis, especially the gonadal biopsy, can be performed to rule out mixed gonadal dysgenesis and developing malignancy [4, 5].

Conclusion

PMDS has an autosomal recessive inheritance. Screening of siblings and second –degree relatives is necessary. Although ultrasonography, CECT and MRI are reported to play a role in locating the mullerian remnants, laparoscopy has a distinctive advantage in diagnosis of PMDS. Familiarity of operating surgeon with this disease condition would increase the chances of correctly diagnosing and appropriately dealing with the mullerian remnants.

Conflict of Interest

Not available.

Financial Support

Not available.

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